GEPHE SUMMARY

Gephebase Gene
agrecan (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^agrecan^#gephebase-summary-title)

GepheID
GP00000072

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Body size (height) (https://www.gephebase.org/search-criteria?/and+Trait=^Body size (height)^#gephebase-summary-title)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens

Ancestral State
Data not curated

Taxonomic Status
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
ACAN

Synonyms
AGC1; SEDK; AGCAN; CSPG1; MSK16; CSPGCP; SSOAOD

String
9606.ENSP00000387356 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000387356)

Sequence Similarities
Belongs to the aggrecan/versican proteoglycan family.

GO - Molecular Function
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0030246 : carbohydrate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030246)
GO:0005201 : extracellular matrix structural constituent (https://www.ebi.ac.uk/QuickGO/term/GO:0005201)

UniProtKB Homo sapiens
P16112 (http://www.uniprot.org/uniprot/P16112)

GenebankID or UniProtKB
M55172 (https://www.ncbi.nlm.nih.gov/nuccore/M55172)

GO:0030021 : extracellular matrix structural constituent conferring compression resistance (https://www.ebi.ac.uk/QuickGO/term/GO:0030021)
GO:0005540 : hyaluronic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005540)

GO - Biological Process

GO:0007155 : cell adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0007155)
GO:0007417 : central nervous system development (https://www.ebi.ac.uk/QuickGO/term/GO:0007417)
GO:0001502 : cartilage condensation (https://www.ebi.ac.uk/QuickGO/term/GO:0001502)
GO:0002063 : chondrocyte development (https://www.ebi.ac.uk/QuickGO/term/GO:0002063)
GO:0030199 : collagen fibril organization (https://www.ebi.ac.uk/QuickGO/term/GO:0030199)
GO:0030198 : extracellular matrix organization (https://www.ebi.ac.uk/QuickGO/term/GO:0030198)
GO:0007507 : heart development (https://www.ebi.ac.uk/QuickGO/term/GO:0007507)
GO:0018146 : keratan sulfate biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0018146)
GO:0042340 : keratan sulfate catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042340)
GO:0030166 : proteoglycan biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0030166)
GO:0006508 : proteolysis (https://www.ebi.ac.uk/QuickGO/term/GO:0006508)
GO:0001501 : skeletal system development (https://www.ebi.ac.uk/QuickGO/term/GO:0001501)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0062023 : collagen-containing extracellular matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0062023)
GO:0005604 : basement membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005604)
GO:0031012 : extracellular matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0031012)
GO:0098982 : GABA-ergic synapse (https://www.ebi.ac.uk/QuickGO/term/GO:0098982)
GO:0098978 : glutamatergic synapse (https://www.ebi.ac.uk/QuickGO/term/GO:0098978)
GO:0005796 : Golgi lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0005796)
GO:0043202 : lysosomal lumen (https://www.ebi.ac.uk/QuickGO/term/GO:0043202)
GO:0098966 : perisynaptic extracellular matrix (https://www.ebi.ac.uk/QuickGO/term/GO:0098966)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

Unknown (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Unknown^#gephebase-summary-title)

Aberration Type

Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)

Molecular Details of the Mutation

unknown

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)

Main Reference

Recent progress in the study of the genetics of height. (2011) (https://pubmed.ncbi.nlm.nih.gov/21340692)

Authors

Lettre G

Abstract

Adult height is a classic polygenic trait of high narrow-sense heritability (h (2) = 0.8). In the late nineteenth to early twentieth century, variation in adult height was used as a model to set the foundation of the fields of statistics and quantitative genetics. More recently, with our increasing knowledge concerning the extent of genetic variation in the human genome, human geneticists have used genome-wide association studies to identify hundreds of loci robustly associated with adult height, providing new insights into human growth and development, and into the architecture of complex human traits. In this review, I highlight the progress made in the last 2 years in understanding how genetic variation controls height variation in humans, including non-Caucasian populations and children.

Additional References

RELATED GEPHE

Related Genes

23 (ADAMTS10, CREBRF, DC-STAMP domain containing 2 (DCST2), DYM, EIF2AK3, FTO, GDF5, GHSR, GPR133, Growth Hormone Receptor (GHR), HMGA2, Insulin-like growth factor receptor 1 (IGF1R), JAZF1, KCNQ1, LCORL, LIN28B, natriuretic peptide precursor type C (NPPC), natriuretic peptide receptor 3 (NPR3), Patched1 (Ptc1), PPAR-delta, TRIP11 (=GMAP-210), SMAD family member 2 (SMAD2), stanniocalcin 2 (STC2)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Body size/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS